A-48: Application of a multiplex polymerase chain reaction system in the detection of dystrophin gene deletion in a preliminary sample of Sri Lankan patients

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Duchenne Muscular Dystrophy (DMD) is a X-linked recessive disease. It is the second most common lethal genetic disorder in humans, affecting 1 in 3500 live male births. Detection of the mutation in an affected subject can greatly enhance the accuracy of prediction of carrier status for female relatives and of subsequent prenatal diagnostic testing. Rapid and efficient methods of screening for deletions by using the polymerase chain reaction (PCR) to analyse frequently deleted exons simultaneously in a multiplex amplification reaction has been reported. After careful clinical evaluation using standard clinical diagnostic criteria for DMD, 23 patients were selected from 22 unrelated Sri Lankan families for screening of deletions using a modified multiplex PCR system.

DNA was extracted from blood samples using standard techniques. PCR primer sequences for the amplification of 18 exons (covering the 2 major 'deletion prone' regions in the gene) in 2 multiplex reactions were custom synthesized. Of the patients studied, 95% had deletions in the 2 major 'deletion prone regions', while one did not show any deletions in these regions. Compared to the traditional method of diagnosis using Southern blot based techniques, PCR is

rapid, cost effective and provides greater resolution, making it the method of choice for routine detection of deletions in the dystrophin gene.